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REFERENCE   1  (bases 1 to 6046)
  AUTHORS   Claes,L., Ceulemans,B., Audenaert,D., Smets,K., Lofgren,A.,
            Del-Favero,J., Ala-Mello,S., Basel-Vanagaite,L., Plecko,B.,
            Raskin,S., Thiry,P., Wolf,N.I., Van Broeckhoven,C. and De Jonghe,P.
  TITLE     De novo SCN1A mutations are a major cause of severe myoclonic
            epilepsy of infancy
  JOURNAL   Hum. Mutat. 21 (6), 615-621 (2003)
  MEDLINE   22637994
   PUBMED   12754708
  REMARK    GeneRIF: de novo mutations in SCN1A are a major cause of isolated
            severe myoclonic epilepsy of infancy (SMEI or Dravet syndrome)
REFERENCE   2  (bases 1 to 6046)
  AUTHORS   Sugawara,T., Tsurubuchi,Y., Fujiwara,T., Mazaki-Miyazaki,E.,
            Nagata,K., Montal,M., Inoue,Y. and Yamakawa,K.
  TITLE     Nav1.1 channels with mutations of severe myoclonic epilepsy in
            infancy display attenuated currents
  JOURNAL   Epilepsy Res. 54 (2-3), 201-207 (2003)
  MEDLINE   22721568
   PUBMED   12837571
  REMARK    GeneRIF: SCN1A channels bearing myoclonic epilepsy (SMEI) nonsense
            and missense mutations show attenuated or barely detectable inward
            sodium currents, indicating that SMEI mutations lead to
            loss-of-function and may contribute to development of SMEI
            phenotypes.
REFERENCE   3  (bases 1 to 6046)
  AUTHORS   Chou,I.C., Peng,C.T., Tsai,F.J., Huang,C.C., Shi,Y.R. and Tsai,C.H.
  TITLE     The lack of association between febrile convulsions and
            polymorphisms in SCN1A
  JOURNAL   Epilepsy Res. 54 (1), 53-57 (2003)
  MEDLINE   22627883
   PUBMED   12742596
  REMARK    GeneRIF: the SCN1A gene might not be one of the susceptibility
            factors for febrile convulsions
REFERENCE   4  (bases 1 to 6046)
  AUTHORS   Gennaro,E., Veggiotti,P., Malacarne,M., Madia,F., Cecconi,M.,
            Cardinali,S., Cassetti,A., Cecconi,I., Bertini,E., Bianchi,A.,
            Gobbi,G. and Zara,F.
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TITLE Familial severe myoclonic epilepsy of infancy: truncation of Nav1.1 and genetic heterogeneity
JOURNAL Epileptic Disord 5 (1), 21-25 (2003)
MEDLINE 22658066
PUBMED 12773292
REMARK GeneRIF: familial severe myoclonic epilepsy of infancy is geneticlly heterogenous for SCN1A (Nav1.1)mutation

REFERENCE 5 (bases 1 to 6046)
AUTHORS Weiss,L.A., Escayg,A., Kearney,J.A., Trudeau,M., MacDonald,B.T., Mori,M., Reichert,J., Buxbaum,J.D. and Meisler,M.H.
TITLE Sodium channels SCN1A, SCN2A and SCN3A in familial autism
JOURNAL Mol. Psychiatry 8 (2), 186-194 (2003)
MEDLINE 22497727
PUBMED 12610651
REMARK GeneRIF: R542Q in SCN1A was observed in one autism family and had previously been identified in a patient with juvenile myoclonic epilepsy

REFERENCE 6 (bases 1 to 6046)
AUTHORS Ohmori,I., Ouchida,M., Ohtsuka,Y., Oka,E. and Shimizu,K.
TITLE Significant correlation of the SCN1A mutations and severe myoclonic epilepsy in infancy
JOURNAL Biochem. Biophys. Res. Commun. 295 (1), 17-23 (2002)
MEDLINE 22078981
PUBMED 12083760
REMARK GeneRIF: Significant correlation of the SCN1A mutations and severe myoclonic epilepsy in infancy

REFERENCE 7 (bases 1 to 6046)
AUTHORS Lossin,C., Wang,D.W., Rhodes,T.H., Vanoye,C.G. and George,A.L. Jr.
TITLE Molecular basis of an inherited epilepsy
JOURNAL Neuron 34 (6), 877-884 (2002)
MEDLINE 22082300
PUBMED 12086636
REMARK GeneRIF: The effects of three mutations in SCN1A have been characterized in cultured mammalian cells as a gain-of-function abnormality causing prolonged membrane depolarization, a plausible underlying biophysical mechanism responsible for inherited epilepsy.

REFERENCE 8 (bases 1 to 6046)
AUTHORS Sugawara,T., Mazaki-Miyazaki,E., Fukushima,K., Shimomura,J., Fujiwara,T., Hamano,S., Inoue,Y. and Yamakawa,K.
TITLE Frequent mutations of SCN1A in severe myoclonic epilepsy in infancy
JOURNAL Neurology 58 (7), 1122-1124 (2002)
MEDLINE 21938587
PUBMED 11940708
REMARK GeneRIF: Three novel frameshift and seven nonsense mutations in the SCN1A gene have been identified in 14 Japanese patients with severe myoclonic epilepsy in infancy.

REFERENCE 9 (bases 1 to 6046)
AUTHORS Escayg,A., MacDonald,B.T., Meisler,M.H., Baulac,S., Huberfeld,G., An-Gourfinkel,I., Brice,A., LeGuern,E., Moulard,B., Chaigne,D., Buresi,C. and Malafosse,A.
TITLE Mutations of SCN1A, encoding a neuronal sodium channel, in two families with GEFS+2
JOURNAL Nat. Genet. 24 (4), 343-345 (2000)
MEDLINE 20206553
PUBMED 10742094

REFERENCE 10 (bases 1 to 6046)
AUTHORS Malo,M.S., Blanchard,B.J., Andresen,J.M., Srivastava,K., Chen,X.N., Li,X., Jabs,E.W., Korenberg,J.R. and Ingram,V.M.
TITLE Localization of a putative human brain sodium channel gene (SCN1A)

to chromosome band 2q24
JOURNAL Cytogenet. Cell Genet. 67 (3), 178-186 (1994)
MEDLINE 94340991
PUBMED 8062593
REFERENCE 11 (bases 1 to 6046)
AUTHORS Lu,C.M., Han,J., Rado,T.A. and Brown,G.B.
TITLE Differential expression of two sodium channel subtypes in human brain
JOURNAL FEBS Lett. 303 (1), 53-58 (1992)
MEDLINE 92275082
PUBMED 1317301
COMMENT PROVISIONAL REFSEQ: This record has not yet been subject to final NCBI review. The reference sequence was derived from AY043484.1.
On Apr 16, 2003 this sequence version replaced gi:21914835.
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